SCHEDULE 2 – THE SERVICES

# A. Service Specifications

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| **Service Specification No:** | 1640 |
| **Service** | Cystinosis (all ages) |
| **Commissioner Lead** | *For local completion* |
| **Provider Lead** | *For local completion* |

## 1. Scope

1.1 **Prescribed Specialised Service**

This service specification covers the provision of management services for patients with Cystinosis, all ages.

1.2 **Description**

Cystinosis is a rare, autosomal recessive genetic disease, which is diagnosed early in childhood, usually before the age of two. It leads to an increase in many parts of the body of the amino acid cystine. This build up causes cystine crystals to form in many organs. These crystals form firstly in the kidneys and the eyes, and later in the muscles, pancreas, thyroid gland and white blood cells.

There are three different types of cystinosis:

1. Nephropathic infantile cystinosis which starts by affecting the kidneys in babies and children under the age of 2 (90-95% of all cases)
2. Nephropathic juvenile cystinosis which starts by affecting the kidneys in young children (3-5% of all cases)
3. Ocular cystinosis which affects the eyes and does not cause problems until later in life (the rarest form with only 1-2% of all cases)

1.3 **How the Service is Differentiated from Services Falling within the Responsibilities of Other Commissioners**

NHS England commissions management services for adults with Cystinosis from Highly Specialist Cystinosis Management centres, including services delivered on an outreach basis delivered as part of a provider network.
Clinical Commissioning Groups (CCGs) commission inpatient care for adults with cystinosis.
Activity is identified via local data flows, which apply to Highly Specialised Cystinosis centres only.

## 2. Care Pathway and Clinical Dependencies

2.1 **Care Pathway**

Please note that access to treatment will be guided by any applicable NHS England national clinical commissioning policies.

The Provider will manage referrals in line with any relevant national or local guidelines or recommendations, and in accordance with the agreed response times. All patients will be managed

by a multi professional team. The model of at least two consultants with a particular interest in cystinosis, supported by other specialists with nursing and other staff with a primary focus on this group of patients reviewed in an out-patient environment with access to in-patient care and monitoring via a multi-disciplinary team (MDT) will be common to all units. The model would complement the care that patients already receive locally.

Referral pathways will be developed between the highly specialised and local renal centres to ensure that both services are made aware of patients as soon as a diagnosis is made and work together to support the care of patients. Written, individualised, transition plan pathways will be used. New management plans will need to be developed that include the following service speciality elements of care:

**Paediatric Cystinosis Services**

Shared care arrangements using nationally standardised protocols will be established with the patient's local renal centres, which in turn will deliver routine children's care with the benefit of easy access and communication with the highly specialised paediatric cystinosis service.

Paediatric services will provide:

- Assistance with the diagnosis of cystinosis
- Help with access to specialised treatment therapies
- Support and advise on management of the renal Fanconi syndrome
- Advise on nutrition, growth and development
- Support for the management of chronic kidney disease
- Support for the initiation of renal replacement therapy
- Assistance with the provision of high quality information and education for patients their families / carers and health care professionals
- Access to a full range of specialised children's services
- Access to specialised clinical research studies
- Assistance with the transition of patients from paediatric to adult care
- Data entry into the appropriate registry
- Review of patients every 6 or 12 months as appropriate

**Adult Cystinosis Services**

Adults with cystinosis require access to many of the same services available in the paediatric centres; however, kidney disease is typically more advanced in adults. Many adults will have established kidney failure and require renal replacement therapy by dialysis or transplantation. However, significant numbers of patients already survive into adulthood with relatively well preserved renal function and continuing renal Fanconi syndrome. Routine care for chronic kidney disease, including renal replacement therapy, will be provided by local nephrology services.

Adult services will focus on all aspects of Cystinosis;

- Continuation of therapy
- Monitoring of therapy
- Management of the multisystem complications associated with Cystinosis.
- Specific advice on treatment
- Access to specialised clinical research studies
- Managing Fanconi syndrome and supporting local services for chronic renal failure and end stage renal disease.

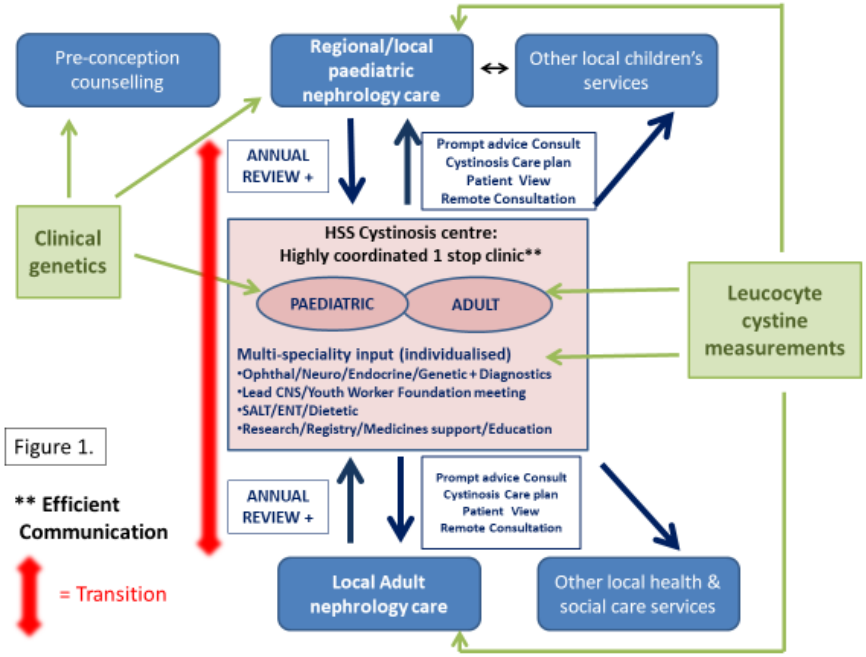


Figure 1.

The Patient Pathway

**Transition Management Plans**

Transition from paediatric to adult services is a major event in a cystinotic patient's life, and there needs to be appropriate and timely systems to facilitate this.

Transition will need to occur between the local paediatric service to the local adult service (e.g. paediatric transplant clinic to adult transplant clinic). In parallel with this, the adult cystinosis service will assume the supporting role from the paediatric cystinosis service and maintain oversight of the process to ensure the specialist support is patient centred and enables patient choice for young adults (age 18-25). This will include those who are in the process of transferring from paediatrics or those who have already transferred. Arrangements for transition to adult services will be considered from the age of 13 and will be completed by the age of 18. In liaison with local providers, the Cystinosis service will develop written transition plans and pathways for each patient.

The transition will involve a period of joint care from paediatric and adult services and it is important for multi-disciplinary teams (MDTs) to offer care that recognises that this group may have additional development needs, including educational and employment.

The following should be put in place:

- Early discussion with the patient and family/carers about transition
- Notification to the adult centre of intention to proceed with transition
- Copies of letters and previous review reports
- At least two clinics will be held at the adult site according to patient needs

The paediatric cystinosis service will be responsible for ensuring all the relevant services have transitioned appropriately.

**Links to an Accredited Laboratory that specialise in Cystinosis relevant diagnostic and management (White Cell Cystine Measurement)**

The Provider will ensure that all relevant diagnostic and management evaluations are carried out within the guidelines and protocols provided by fully accredited laboratories.

The genetic testing associated with this service should be undertaken in line with the NHS England National Genomic Testing Directory for rare and inherited disease, which is on the NHS England website at: https://www.england.nhs.uk/publication/national-genomic-test-directories/.
Genetic tests are delivered by the Genomic Laboratory Hubs, details of which are on the NHS England website at: https://www.england.nhs.uk/genomics/genomic-laboratory-hubs/.

**Care Co-ordinator/Clinical Nurse Specialist**
A designated, named clinician should be identified as care co-ordinator for each patient who is responsible for ensuring the delivery of coordinated care according to the hub and spoke arrangement for that patient.

**Named Consultant**
Each patient should have a named Consultant at the hub centre who is accountable for the delivery of hub services for that patient.

**Psychology Services**
Patients with cystinosis frequently have a variety of psychological problems related to their disease and its complications. This can significantly affect their quality of life and prevent them from maximising their educational potential, gaining employment, integrating into society, and developing significant relationships. While many of these problems are common to patients with chronic long term conditions and renal failure, some will be directly related to their cystinosis. Therefore, the Provider will ensure that patients have access to appropriate psychology services and that the initial assessment will be by professional who have knowledge of cystinosis and its complications.

**Patient Support Groups**
Living with a rare, chronic, multisystem, disease such as cystinosis presents challenges throughout the patient journey from diagnosis through to adulthood. Education, in a form that is understandable and meaningful to all patients and their families/carers (as well as others that they may rely on such as teachers and employers), is an important ingredient in meeting this challenge. Patient support groups such as the Cystinosis Foundation UK, play an important role in providing this education. Working closely with the Cystinosis services they can help produce educational material, participate in face-to-face educational activities and peer-to-peer meetings and generally increase awareness of the disease and the issues that patients experience. Providers will ensure relevant patient organisations are involved in all relevant educational initiatives and activities. In addition, Providers will be required to ensure that relevant information, together with appropriate contact details and sources of further information, such as web links, is available at all clinics.

**Information System**
The Provider will utilise information systems which meet the requirements to submit data to the UK Renal Registry of the Renal Association and its Rare Disease Registry (RADAR) for comparative audit purposes, and other local requirements set out by the commissioner of the service. Clinical audit information should be made available to patients in an accessible format.

The electronic record in the renal information systems will enable data fields in the registry to be completed.

**Technology & Facilities**
The Provider will need to develop systems to support the cystinosis centre approach and technology (equipment) and facilities that will promote improved care for patients with cystinosis. For example, the Provider may need to use Telecare / Telemedicine technology to coordinate pathways with locally provided services.

Patients and their families will have access to Patient View. It is anticipated that their comprehensive care plan will be uploaded to Patient View and updated as needed thereby creating an “electronic patient passport”.

2.2 **Interdependence with other Services**

The multisystem nature of cystinosis means that as well as life-long kidney care, there must be access to other specialists with specific expertise in the non-renal complications of cystinosis.

Providers will ensure that access in a "one stop" multidisciplinary clinic, annually or more frequently if indicated / needed, offering clinical consultations and all necessary investigations. Additional access to those specialists will be provided as required between annual reviews.

Each patient's comprehensive care plan will detail their requirements and the clinics will be responsive to changing needs.

The required expertise needs to be available as part of the MDT as specified:

| Service / Area of expertise | Paediatrics | Adults |
|---|---|---|
| Clinical Nurse Specialist | Essential | Essential |
| Clinical genetics – Diagnostic | Essential | As necessary |
| Pregnancy / Counselling | As necessary | As necessary |
| Pharmacy - overseeing access to specialised therapies | Essential | Essential |
| Nutritional Support | Essential | As necessary |
| Growth & development | Essential | - |
| Ophthalmic | Essential | Essential |
| Gastro-intestinal | As necessary | As necessary |
| Neurological & Neuromuscular (include respiratory function) | Essential | Essential |
| Physiotherapy | As necessary | Essential |
| Speech & Language therapy | Essential | Essential |
| Endocrine | Essential | As necessary |
| Cardiac | As necessary | As necessary |
| Reproductive health | - | Non-core |
| Psychological support | Essential | Essential |

**Nephrology Services**

Providers should develop detailed care pathways across both elements of paediatric and adult nephrology care for diagnosis and monitoring of relevant care processes associated with the disease, in line with national / local evidence recommendations.

Paediatric nephrology services should incorporate the following elements into the patient pathway:

- Support and advice for diagnosis
- Support and advice for laboratory assessment & confirmatory genetic mutation testing
- Support and advice for early management of renal care
- Support and advice for fluid and electrolyte assessment & management, especially of renal Fanconi syndrome
- Support and advice for CKD assessment & management
- Support and advice for initiation of Renal Replacement Therapy
- Support and advice for coordination of cystinosis specific tests including measurement of cystine levels

**Transition management of patients (covered in previous sections.)**

Adult Nephrology Services will need to provide a continuation of the paediatric nephrology pathway as well as:

**Patients not on renal replacement therapy:**

- Support and advice for management of continuing fluid and electrolyte problems, especially of renal Fanconi syndrome
- Support advice and coordination for management of CKD

- Support and advice for monitoring & continued assessment discussion / referral for dialysis and transplantation (to local renal centre; dialysis and transplantation delivery is outside of this specification)
- Support and advice for ongoing cystinosis-specific therapy and management
- Accessing Laboratory tests and results:
- Routine laboratory assessments
- Laboratory tests specific to the management of CKD and its complications
- Laboratory tests specific to cystinosis

**Transplant and dialysis patients:**

Unless living within the catchment of the highly specialised centre, patients with transplants and on dialysis will be managed by their local renal centre. However, the cystinosis service will offer support and advice from physicians experienced in issues specific to cystinosis, as appropriate from the list above, for example fluid, electrolyte and blood pressure management. The HSS will offer support and advice for management plans regarding choice & access to various forms of renal replacement coordination.

**Ophthalmology Services**

Ocular involvement in cystinosis is the first major complication to develop and without pro-active management can lead to photophobia, blepharospasm, and ultimately to severe pain and impaired vision. In addition, there is a known risk of raised intracranial pressure in nephropathic cystinosis, which may present first with papilloedema. Providers will need to provide an integrated Ophthalmology pathway that includes elements of the following:

- Annual review
- Coordination of regular eye screening and management

**Facilities required**

A fully equipped ophthalmology suite with equipment to detect and manage the following:

- Crystal deposition in the cornea detected by slit lamp biomicroscopy or hand held slit lamp biomicroscopy in children less than 4 years of age.
- If treatment is delayed or not effective the following complication will need to be detected and managed
- Photophobia
- Blepharospasm
- Superficial punctate keratopathy and ocular surface disease
- Corneal erosions
- Retinal crystal deposition
- Raised intracranial pressure which can present in children or adults and requires monitoring in conjunction with neurologists and paediatricians. If raised ICP is diagnosed monitoring of this can be done by local ophthalmologists as this may require frequent follow up examinations
- Additional equipment required includes optical coherence tomography for optic nerve and macula including wide angle fundal digital imaging

In older patients the following less common conditions occur and the facilities should allow accurate detection and management:

- Filamentary and band keratopathy
- Corneal neo-vascularisation
- Retinopathy

- Glaucoma
- Retinal and optic nerve degeneration

Laboratory links

- No specific laboratory tests are needed.
- However, some of the complication can have other causes and so may need access to the usual laboratory links that a specialised ophthalmology service might access such as retinal angiography and specialist optometry services.

**Neurology Services**

For adult patients with cystinosis, access to support from a Neurologist with expertise in the care of patients with cystinosis is essential. Cystine accumulates in cells throughout the body and causes progressive damage to multiple organs, including widespread muscle weakness and the brain.

In the paediatric population there is evidence of neuro-cognitive involvement.

- Patients should have access to appropriately experienced paediatric neurologist.
- Cognitive function needs to be tested at regular intervals and this will need access to appropriate paediatric psychology services.
- Access to appropriate neuro-imaging (MRI) will be required.

In adults the problems can be sub-divided as follows:

- Neuro-cognition
  - Same issues as in children and will need following; possibly with help of psychologists.
  - Also included raised intra-cranial pressure (ophthalmic review and cerebrovascular events
    - links to neuro-imaging and to neuro-surgery
- Myopathy related issues; this will need early detection and there should be access to a neurologist experienced in the diagnosis and management of muscle diseases. The following facilities may be needed:
  - Access to a laboratory able to process specimens routinely requested by neurologists managing muscle disease (to exclude other causes of myopathy)
  - Support of a physiotherapist to perform tests such as manual dexterity and walking ability—e.g. 6 minute walk test. These tests will require space and some appropriate simple equipment.
  - Muscle power in the appropriate muscle groups will be recorded by use of the MRC muscle power grading or similar.

Cystinosis can involve the muscles that are used to breathe. This can cause problems leading to respiratory failure, sleep apnoea, and breathlessness. To diagnose these serious problems patients will need access to either a neurologist with a special interest in neuro-respiratory problems or another physician (such as a chest physician) with similar expertise. In order to properly assess such patients the following tests should be readily available:

- Routine spirometry and access to a pulmonary function laboratory is required for:
- Random blood gases in an OPD setting (the OPD needs to be near a lab or other facility that can measure gases)
- Ability to measure mouth inspiratory and expiratory pressures or sneeze inspiratory and expiratory pressures
- Access to assisted ventilation expert advice

Providers will need to offer patients with cystinosis access to a neurologist experienced in the management of neurological problems associated with cystinosis that may include;

- Assessments for Muscle Disease
- Co-ordinated interventions for patients with signs / symptoms of Neurological complications

**Speech and Language Services**

Speech and language care for individuals with cystinosis may be linked with neuromotor dysfunction, that make feeding and swallowing difficult for some. Hence speech and language services are essential. Providers will have to develop an integrated pathway that incorporates the following speech and language service elements;

- Review and coordination with locally signposted services
- Access to speech and language assessments e.g. videofluoroscopy
- Co-ordinated action plans for patients that require interventions

**Nutrition and Dietetics Services**

Nutritional issues occur throughout the life of a cystinotic patient. Providers will need to develop a pathway that incorporates and integrates nutritional and dietetics services into their cystinosis service pathway. Patients will be seen by the dietician as required. In addition, patients will have access to telephone advice and literature as clinically required. The pathway may include elements of the following;

- Regular reviews
- Access to assessments
- Access to procedures such as enteral feeding

In early life it is important that patients receive adequate nutrition as well as the supplements that will require for their Fanconi syndrome.

- Access to a dietician experienced in management of such problems
- Access to nutritional support procedures such as enteral feeding etc.
- Further laboratory monitoring may be needed

With the development of renal impairment and then renal failure, dietary intervention from an appropriately experienced dietician will be required. This will also be needed when/if renal replacement therapy is initiated but will be delivered locally to the patient with close coordination with dietician.

Diabetes occurs in approximately 25% cases and may require the following:

- Dietary advice from an appropriately experienced dietician
- Dietetic advice may also be needed for help in management of other problems such as abdominal pain.

**Endocrinology Services**

All patients with cystinosis should have access to expert comprehensive endocrine care. Providers will need to develop a service pathway that incorporates & integrates all aspects of endocrine care relevant to cystinosis and its complications.

**Growth & Development (monitoring & interventions)-Paediatric Centres**

- Growth can be restricted in cystinosis
- Charts to record height and weight and head circumference should be available and measurements taken regularly
- Growth hormone may be indicated as per accepted guidelines.

**Diabetes (Assessment, Monitoring & interventions)**

This occurs in many patients although good control of cystine levels can slow the onset to this complication. Patients should have access to specialist diabetes clinics. It is important that the self-management strategies that are taught to diabetics are adapted to the needs of a person living with cystinosis. Diabetes care will predominantly be provided locally

**Thyroid**
Thyroid complications may occur in a significant number of patients. Thyroxine replacement should be provided and monitored locally with advice from the specialist centre.

**Bone (Linked to Renal Section)**
In paediatric practice bone problems are caused by the loss of calcium and phosphate in the urine along with other ions as a result of the Fanconi syndrome together with a decreased activation of vitamin D. In older patients or those with advanced renal disease or receiving dialysis the situation is more complex and is known as renal osteodystrophy. In the case of renal osteodystrophy management will be in accordance with published guidelines.

- Access to a laboratory able to measure PTH and vit D as well as routine tests will be required
- More complex renal bone disease will be managed by renal physicians and others experienced in such problems and they may need access to further more specialised investigations.

**Sexual function (Linked to Infertility section below)**

- Woman may have delayed menarche, but this rarely needs any specialist advice
- Men on the other hand are infertile and may also testosterone deficient. Identification of testosterone deficiency and signposting of advice on management is key. Testosterone replacement monitoring may be managed locally with collaboration with specialist centre
- Access to assisted conception expertise is required for a proportion of men requesting this service. Expert advice and signposting to contraception services for women).
- Expert pre-conception counselling for women contemplating pregnancy should be available and access to experienced high risk obstetric antenatal services is required for these women

**Dermatology Services**
Cystinosis patients are fair skinned and need advice on skin care. Access to dermatology advice may be needed. Patients after kidney transplantation have an increased incidence of skin cancer; hence transplanted patients with cystinosis may need to be screened for cancer. The Provider will therefore be required to ensure that there is coordination with local dermatology services. These services can be delivered locally

**Genetics & Laboratory Investigation Services (Paediatric Centres)**
As cystinosis is a genetic disease both patients and parents will need appropriate genetic advice The Provider should identify and incorporate the following into their pathways:

- Specific lab tests (measurement of white cell cystine)
- Genetic diagnosis
- Genetic Counselling; as cystinosis is a genetic disease both patients and parents will need access to appropriate genetic advice either from an appropriately trained counsellor and/or a geneticist.

**Fertility treatment & Special Obstetric Advice Service**
Women with cystinosis are fertile; however they will need access to obstetricians experienced in the management of medical problems in pregnant patients. The Cystinosis services must work with local obstetric services to incorporate the following into their service pathway:

- Drug therapy adjustments during pregnancy
- Renal transplant guidelines during pregnancy

Men are infertile and may also be testosterone deficient. The Provider may rarely need to incorporate the following into their service:

- Advice of reproductive possibilities

**Pharmacy Services**
The Provider will ensure that medications required are prescribed and dispensed appropriately. Specific paediatric pharmacist expertise is required for children who may require 5-8 medications each several times a day, in liquid or palatable formulations (some of which may be unlicensed or off-label).

**Medicines Management**
The Provider is expected to have in place a medicines management procedure.

The Provider shall comply with the higher standards of (i) National Minimum Standards and (ii) the highest available Clinical Negligence Scheme for Trusts (CNST) Standard in relation to medicines management as updated from time to time. Patients should receive a regular scheduled formal medicines management review and this should be done routinely whenever a patient changes clinical locations for their care. The use of cysteamine can be associated with certain problems that can be addressed by a pharmacist experienced in cystinosis and its treatment.

## 3. Population Covered and Population Needs

### 3.1 Population Covered by this Specification
The scope of this service is all adults and children in England with a diagnosis of cystinosis. The service outlined in this specification is for patients ordinarily resident in England.

### 3.2 Any acceptance and exclusion criteria and thresholds
There are no exclusions. Access to these services will be offered to all adults and children in England with a diagnosis or suspected diagnosis of cystinosis.

### 3.2 Population Needs
Cystinosis is a recessively inherited metabolic disease characterized by an accumulation of cystine within the lysosome compartment of cells, causing damage to many organs and tissues, including the kidneys and eyes. It is estimated that cystinosis occurs in somewhere between 1 in 100,000 to 1 in 200,000 live births. There are 2 or 3 new cases of cystinosis diagnosed each year in the UK. The disease is pan-ethnic with no particular geographical distribution within the UK. However, as a recessive trait, the disease is more common in communities with a high incidence of consanguinity.

Three clinical forms have been described: [Nephropathic refers to cases that start with kidney involvement]

- Nephropathic infantile; the severest form accounting for about 90-95% of all cases.
- Nephropathic juvenile; accounting for about 3-5% of cases.
- Ocular; the late onset form that affects the eyes, although occasional cases have had some mild kidney involvement: accounting for around 2-3% of cases.

Effective management of patients with cystinosis is complex and involves the provision of comprehensive care by a team of health care professionals with diverse and complementary skills.

Advances in medical treatment have led to improved long-term survival of children with cystinosis. Whereas nearly all children born in the 1970s died in childhood, now almost half of the UK cystinosis patient population are adults.

Services will provide world-class coordinated care to those needing the most specialist treatment echoing the UK Department of Health Strategy for Rare Diseases. (https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/260562/UK_Strategy_for_Rare_Diseases.pdf).

These centres would act as hubs in a new connected system of care including specialist services, local hospitals and primary and community care services providing consistently excellent services and delivering most care close to patients' homes. Expert hubs would help to delay and/or prevent severe complications such as hypothyroidism, diabetes mellitus, established chronic kidney disease and respiratory disease & impaired swallowing due to profound muscle weakness.

Even with adequate treatment all patients will, at some time, develop established renal failure and require dialysis and/or renal transplantation. In addition, nearly all patients will develop corneal disease and other eye problems as well as a significant number developing diabetes and hypothyroidism. Other complications include male infertility, bone disease, neurocognitive dysfunction, myopathy, respiratory compromise and difficulties in swallowing.

Therefore, appropriate care requires a multi-disciplinary and multi-professional approach to overall management.

This service model has been developed following a period of extensive consultation with the UK nephrology community, patients and carers, including members of a UK Renal Association Rare Disease Cystinosis Study Group of English Nephrologists (paediatric and adult) from Units with considerable experience in the management of patients with cystinosis in association with the Cystinosis Foundation (UK) representing patients & with Pharma and those engaged in R&D in respect of cystinosis.

### 3.3 Expected Significant Future Demographic Changes

It is likely that the incidence of live births with cystinosis will remain static. It is possible that increased awareness of the condition will result in more cases identified year on year into paediatric centres. With improvements in care and reduced mortality, there will be a predictable gradual increase in the adult population.

### 3.4 Evidence Base

Individuals living with rare diseases often need expertise from a number of specialists, making a multidisciplinary and coordinated team approach important and for patients with cystinosis this is definitely true.

Well-coordinated care is essential when several specialists and hospital departments are involved in a patient's care. It is not the best use of time or resources if patients have to visit different departments at the same hospital on different days, particularly if the hospital is not close to their home. Problems can also occur if interactions between different treatments are not properly managed.

Due to the large number of specialities involved in the care for patients living with cystinosis, a large number of Clinical Guidelines and Best Practice Protocols will need to be amalgamated and adopted by the Cystinosis centres. The following is a list of some such references to guidance:

- The National Service Framework (NSF) for Renal Services (Department of Health, 2004/5) https://www.gov.uk/government/publications/national-service-framework-kidney-disease
- NICE Guidance CG182: Chronic kidney disease in adults https://www.nice.org.uk/guidance/cg182
- NICE Guidance NG107 Renal Replacement therapy and conservative management https://www.nice.org.uk/guidance/ng107
- UK RENAL Association: Information on Rare Renal Diseases. http://rarerenal.org/rare-disease-groups/cystinosis-rdg/
- UK Strategy for Rare Disease https://www.england.nhs.uk/wp-content/uploads/2018/01/implementation-plan-uk-strategy-for-rare-diseases.pdf

- End of Life Care in Advanced Kidney Disease http://www.ncpc.org.uk/sites/default/files/EndOfLifeCareInAdvancedKidneyDisease.pdf
- Nephropathic cystinosis: an international consensus document. http://europepmc.org/articles/pmc4158338
- KDIGO Guidelines https://kdigo.org/guidelines/
- The Renal Association: Clinical Practice Guidelines https://renal.org/guidelines/
- British Transplantation Society Guidelines https://bts.org.uk/guidelines-standards/
- Standards for the investigation of respiratory complications of muscle disease [Qaseem A et al. Management of Obstructive Sleep Apnoea in Adults: A Clinical Practice Guideline from the American College of Physicians. Ann Intern Med 2013: 159:471-483] and a standard scoring system [Epworth sleepiness scale]
- Garber J R et al ATA/AACE guidelines in hypothyroidism in adults. Endocr Pract 2012 18(6): 988-1028
- NICE Guidance NG17. Type 1 Diabetes https://www.nice.org.uk/guidance/ng17
- NICE Guidance NG28. The Management of type 2 diabetes https://www.nice.org.uk/guidance/ng28
- NICE Guidance NG 18 diabetes in children and young people https://www.nice.org.uk/guidance/ng18

## 4. Outcomes and Applicable Quality Standards

### 4.1 Quality Statement – Aim of Service

The service model will aim to balance the needs of patients and their families / carers to receive routine care that might involve frequent visits to local centres (hospitals / spokes) with the need for regular but less frequent visits (e.g. 6-12 monthly) to highly specialised, multi-disciplinary, and multi-professional clinics with a critical mass of expertise and experience of care for patients with cystinosis across all age groups. The model will apply equally to adult and children's services, although the role of the centre may vary for the two groups of patients.

The model stresses the importance of proper transition (https://www.nice.org.uk/guidance/ng43). There needs to be well organised, multiprofessional and multidisciplinary pathways for individual patients to transfer from paediatric to adult care. This should involve all elements of a patient's care in a holistic manner. It is unrealistic to provide full transitional care to the hub centre. Coordination is required with the focus of transitional care will be from regional paediatric care to local adult renal unit

Services will be patient-centred offering holistic, safe, effective, evidence-based therapies in appropriate care settings. Patients will be supported to make informed choices regarding treatment options and in managing their condition so as to achieve their goals, their best outcomes and their best quality of life.

A service should provide each patient with a minimum of an annual multi-disciplinary review at the highly specialised centre which will endeavour to deliver and perform this via a "one-stop" service. The vision for this approach means that patients would be seen by relevant specialists on the same day, and likely to be in the same location to minimise appointments and maximise the interaction between all parties. Hubs will ensure coordination and interpretation of investigations and make recommendations across the patient pathway, arranging all appointments for relevant assessment investigations in advance. The service will compliment and support the care that is delivered locally and avoid repetition of investigations unnecessarily.

Investigations required at annual review (Adults)

- Laboratory tests: WBC Cystine levels
- Full biochemical and CKD/transplant related routine testing

- Spirometry
- SALT assessment
- Ophthalmic review
- Tests of thyroid function, HBA1c, testosterone, LH, FSH and oestradiol

Centres provide a full report to all concerned (including the patient and, where relevant, their families or carers) within 14 working days. Supplementary letters will need to be sent if certain test results are not immediately available.

Centres will offer comprehensive assessments and provide recommendations for care. This will include the following:

- Promote equity of access allowing everyone with cystinosis to follow a comprehensive disease care pathway, providing high quality, holistic services for every individual through integrated personal care plans.
- Facilitate a patient centered, coordinated approach to investigation, treatment, specialist healthcare and social care support, where necessary. This will take into account the needs of patients, their families and others who provide essential support.
- Deliver evidence-based management developed through the best use of educational resources that are easily accessible by patients and professionals.
- Support specialised clinical centres to provide expert, high quality, clinical care and expertise to patients their families and carers, through a multiprofessional health care team.
- Promote excellence in research to advance our understanding of cystinosis and its treatment.
- Deliver rapid and effective translation of advances in the understanding of cystinosis into clinical care by creating appropriate infrastructure, care pathways, and clinical competences.
- Deliver effective interventions and support to patients and their families quickly, equitably, and sustainably.
- Promote collaborative working between the NHS, research communities, academia, and industry wherever possible to facilitate better understanding of cystinosis and its management. Promote England as a first choice location for research into rare disease as a leader, partner, and collaborator.
- Support education and training programmes that enable health and social care professionals to better identify rare diseases helping deliver faster diagnosis and access to treatment pathways for patients.
- Develop a multidisciplinary approach by the centre with sustainability ensured by the presence of at least two consultant physicians as leads for the service.
- The service will collect data and use it for audit purposes, sharing results and action plans with all concerned with the pathways.
- To ensure efficiency and value for money by optimising health resources.
- Working in partnership with patient organisations to support their work.
- Provide online access to investigation results, clinical letters and patient information through Patient View or equivalent.

The tenets of achieving the best outcomes for patients with cystinosis are early diagnosis, early and sustained optimal medical management together with multidisciplinary input. In particular, the early optimal administration of cysteamine has been shown to delay the onset of complications such as renal failure, thyroid disease and diabetes.

The Provider will need to have close links to a metabolic laboratory responsible for the measurement of white blood cell cystine. Nationally there is wide variation in the time taken to obtain these results due to batching (up to 2 months). Establishing a hub and spoke model will allow planned batching and more timely assays (with the aim for results within 1 week).

**NHS Outcomes Framework Domains**

| | | |
|---|---|---|
| **Domain 1** | **Preventing people from dying prematurely** | ✓ |
| **Domain 2** | **Enhancing quality of life for people with long-term conditions** | ✓ |
| **Domain 3** | **Helping people to recover from episodes of ill-health or following injury** | ✓ |
| **Domain 4** | **Ensuring people have a positive experience of care** | ✓ |
| **Domain 5** | **Treating and caring for people in safe environment and protecting them from avoidable harm** | ✓ |

Patient Reported Outcome and Experience Measures should be prioritised and should be the principle barometer of improved quality of life and experience for both patients and their parents/carers.

**National Renal Dataset**

The Provider will ensure that the required patient activity and outcomes data is provided in accordance with the requirements of the UK Renal Registry and RADAR. The National Renal Dataset has been approved as a Full Operational Information Standard by the Information Standards Board for Health and Social Care. The dataset is mandated for collection by the Department of Health, https://www.datadictionary.nhs.uk/.

Each Hub Centre will be expected to enter data into the RADAR cystinosis registry that is being developed along the lines of the French version RADICO (a copy is included in appendix two).

The Provider will also ensure that the required patient activity and outcomes data is provided in accordance with the requirements of the UK Renal Registry: https://www.renalreg.org/data/

**Patient View System**

The provider will ensure that health records relating to a patient's treatment are entered in to the national Patient View System for those patients who consent to the system being used https://www.patientview.org/#/.

Work will need to be done to ensure that all clinicians will be able to access the IT systems mentioned above.

**4.2 Quality standards specific to the service using the following template:**

**Indicators Include:**

| Number | Indicator | Data Source | Outcome Framework Domain | CQC Key question |
|---|---|---|---|---|
| **Clinical Outcomes** | | | | |
| 101 | Proportion of adult patients reviewed at six monthly intervals | Self declaration | 2,4 | Well led, effective, caring |
| 102 | Proportion of adult patients reviewed at least annually | Self declaration | 2,4 | Well led, effective, caring |
| 103 | Proportion of paediatric patients reviewed at six monthly intervals | Self declaration | 2,4 | Well led, effective, caring |
| 104 | Proportion of paediatric patients reviewed at least annually | Self declaration | 2,4 | Well led, effective, caring |

| | | | | |
|---|---|---|---|---|
| 105 | Proportion of patients with end stage kidney disease where transplantation is undertaken | Self declaration | 2,4 | Well led, effective, caring |
| 106 | Proportion of patient details emailed to key stakeholders within two weeks of the patient appointment | Self declaration | 2,4 | Well led, effective, caring |
| 107 | Proportion of patients recruited to a clinical trial | Self declaration | 2,4 | Well led, effective, caring |
| 108 | Proportion of admissions that were unplanned | Self declaration | 2,4 | Well led, effective, caring |
| **Patient Experience** | | | | |
| 201 | A patient experience exercise is undertaken at least two yearly. | Self declaration | 2,4 | Responsive |
| 202 | There is information for parents and carers. | Self declaration | 2,4 | Effective, caring |
| 203 | Patients are provided with an alert card | Self declaration | 2,4 | Effective, caring |
| 204 | Patients are given the details of the cystinosis team member co-ordinating their care | Self declaration | 2,4 | Effective, caring |
| **Structure and Process** | | | | |
| 301 | The provider has a cystinosis team as outlined in the service specification. | Self declaration | 2,3,4,5 | Well led, effective, caring, safe |
| 302 | There are one stop multi-disciplinary clinics available | Self declaration | 2,3,4,5 | Well led, effective, caring, safe |
| 303 | There are transition pathways in place as defined within the service specification. | Self declaration | 1,2,3,4,5 | Safe, effective, caring |
| 304 | Results of a patient's consultation will be uploaded to “Patient View”. | Self declaration | 2,4 | Effective, caring |
| 305 | There are agreed patient pathways as per the service specification. | Self declaration | 1,2,3,4,5 | Safe, effective, caring |
| 306 | There are clinical guidelines as per the service specification | Self declaration | 1,2,3,5 | Safe, effective, caring |
| 307 | There is a process in place that enables communication with referring teams and outreach centres and other hubs | Self declaration | 2,4 | Effective, caring |
| 308 | The service submits data to RaDaR | Self declaration | 2,4 | Safe, effective |
| 309 | The provider reviews annually its contribution to research, trials and other well designed studies | Self declaration | 2,4 | Safe, effective |

**Detailed definitions of indicators, setting out how they will be measured, is included in schedule 6.**

**4.3 Commissioned providers are required to participate in annual quality assurance and collect and submit data to support the assessment of compliance with the service specification as set out in Schedule 4A-C**

**4.4 Applicable CQUIN goals are set out in Schedule 4D**

To be agreed with the Commissioner

## 5. Applicable Service Standards

**5.1 Applicable Obligatory National Standards** *(e.g. NICE Technical Appraisal Guidance, mandatory accreditation requirements)*

**Applicable national standards e.g. NICE**
The Provider is expected to comply with all relevant legislative provisions and the Care Standards Act (2000), and to provide services in accordance with regulations as defined by, but not limited to, the following authorities and organisations which may change over time:

Each centre must assure that: -

i. All practitioners participate in continuous professional development and networking
ii. Patient outcome data is recorded and audited across the service
iii. All centres must participate in the national audit commissioned by NHS England.

Audit meetings should address:

- Clinical performance and outcome
- Process-related indicators e.g. efficiency of the assessment process, prescribing policy, bed provision and occupancy, outpatient follow-up etc.
- Stakeholder satisfaction, including feedback from patients, their families, referring clinician and GPs.

**5.2 Other Applicable National Standards to be met by Commissioned Providers**

**Applicable standards set out in Guidance and/or issued by a competent body (e.g. Royal Colleges)**

- Care Quality Commission and any successor organisations; and
- All applicable law on Health and Safety at work
- Anti-discrimination and equal opportunities legislation
- General Medical Council

**5.3 Other Applicable Local Standards**

**Professional bodies with an interest and national guidance**

- Renal Association and UK Renal Registry
- BAPN
- BPNA
- BSPED
- Royal College of Ophthalmologists
- British Renal Society
- British Transplantation Society including all relevant clinical practice guidelines
- National and local health service bodies and relevant local government authorities
- Regional Renal Clinical Networks (where exist)
- All relevant NICE clinical guidance
- NHS Employment Check Standards

CNST General Clinical Risk Management standard appropriate to the service being delivered:

- National Service Framework for Renal Services
- Royal College of Physicians Clinical Standards for Renal Services

## 6. Designated Providers (if applicable)

To be agreed through a procurement process. It is proposed that 3 adult and 3 paediatric hubs be commissioned. 3 geographically opportune clinics in England is likely to be best in terms of patient travel time and access to multidisciplinary resources.

## 7. Abbreviation and Acronyms Explained

The following abbreviations and acronyms have been used in this document:

*<please list, explaining each abbreviation and acronym)*

Date published: *<insert publication date>*